

Title of the article:

“An analysis of the histopathology, immunohistochemistry and clinical features of rhabdomyosarcoma and a pilot study to correlate the immunomorphology of alveolar and unclassified rhabdomyosarcoma with PAX3/7-FOXO1(FKHR) translocation by PCR”.

Department : Department of General Pathology

Name of the candidate : S. Radhika

Degree and subject : M.D. Pathology

Name of the guide : Dr. Anne Jennifer Prabhu, M.D. Pathology

Objectives:

To analyse the clinicopathological and immunohistochemical features of Rhabdomyosarcomas (RMS) in adult and pediatric population and to identify PAX3/7-FOXO1 translocation in alveolar and unclassifiable RMS.

Methods:

A prospective and retrospective analysis of immunomorphological features, PAX3/7-FOXO1 translocation and clinical outcome for 71 subjects was done from Jan 2012 to Mar 2016. Molecular analysis for PAX3/7-FOXO1 translocation was done by PCR.

Pearson's Chi square test, Kaplan Meier's test and univariate & multivariate analyses were used for analysis.

Results:

- Patient age ranged from 2 months-65 years, predominantly males with equal distribution of embryonal and alveolar (36.62%) subtypes.

- Head & neck (28.17%) was the common site.
- Most of them were >5cm and belonged to stage 4, lymphnode being the commonest metastatic site.
- Tumour necrosis and reparative changes were the common postchemotherapy changes. Strong immunostaining for myogenin was seen in ARMS.
- Metastasis and recurrence were common in ARMS, the unfavourable sites with higher rate of metastasis.
- The overall survival rate was 93.7% with a median survival duration of 86 days (14-158 days) during the followup period of 2 days to 40 months. The overall DFS was 28 days (77-496 days). The DFS was 312 days (132-491 days) for alveolar RMS. The median DFS was 83 days (20.5-145.4 days) for 12 cases which had recurrence.
- Translocation (PAX3/FOXO1) was found only in one typical ARMS.

Conclusions:

- Patients were predominantly males, <10 years.
- Strong(3+) myogenin staining was found in ARMS, which also showed increased risk of metastasis and recurrence.
- Unfavourable site predicted poor outcome.
- PAX3/FOXO1 translocation in only one case could be explained by a possibly low frequency of translocation in the Indian population.

Key-words: Rhabdomyosarcoma, PCR, alveolar rhabdomyosarcoma, Desmin, Myogenin, PAX3/7-FOXO1, ARMS- alveolar rhabdomyosarcoma.